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IMAGES IN CARDIOLOGY

Multimodality Imaging of Cardiac Involvement in Neurofibromatosis

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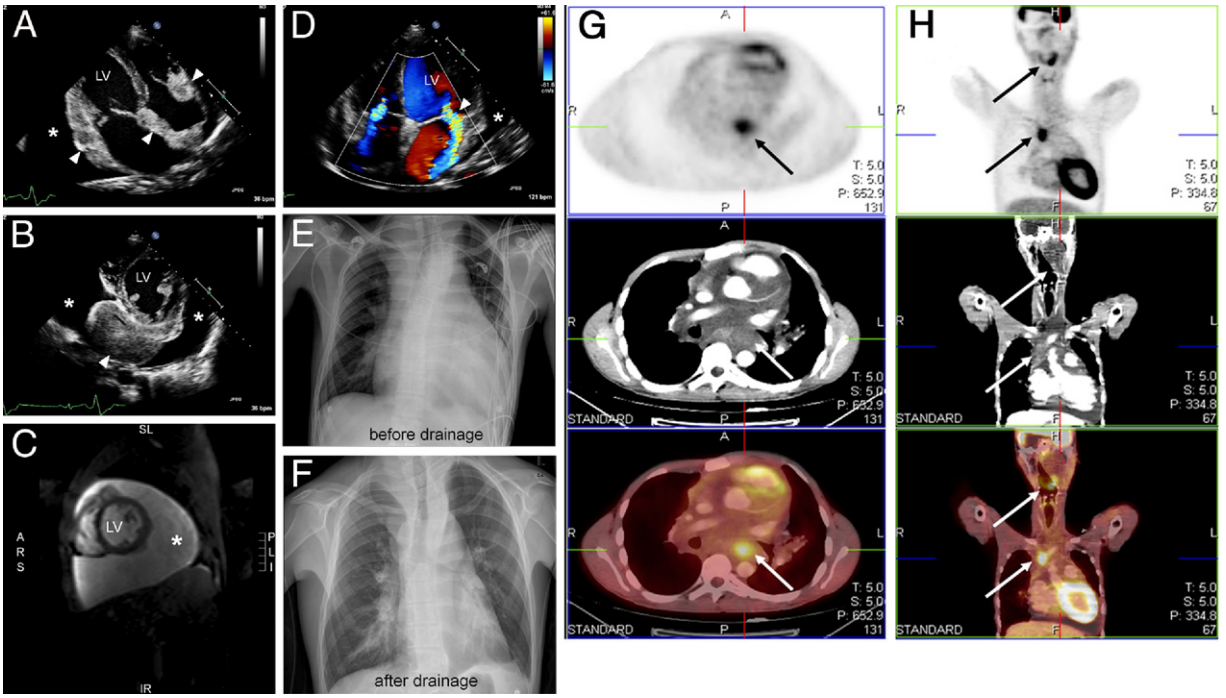

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A 21-year-old man with neurofibromatosis type 1 neglected follow-up during a decade and presented with progressive exertional dyspnea and thoracic tightness. Echocardiography revealed an infiltrative mass (**A, arrowheads,** Online Video 1) originating from the basal ventricular segments, the lateral sides of both atria, and the interatrial septum, forming a retrocardiac mass (**B, arrowhead**, Online Video 2). The extent of impressive pericardial effusion **(asterisk)** was best shown by cardiac magnetic resonance **(C)**. Severe mitral regurgitation secondary to ventricular remodeling and restrictive leaflet motion was present (**D, arrowhead**, Online Video 3). Laparoscopic-guided biopsy of the retroperitoneal mass that extended in the mediastinum and neck showed neurofibroma. Sixteen hundred milliliters of pericardial effusion with benign histology was drained percutaneously, providing full symptomatic relief **(E, F)**. Multiple hot spots on positron emission tomography, however, strongly suggested malignant reconversion, implicating an adverse outcome (**G, H, arrows**, Online Video 4). Cardiac involvement of neurofibromatosis is extremely rare. Further invasive testing for histological characterization of the hot spots is being considered. LV = left ventricle.